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375 Cystic fibrosis, apple peel disease and normal neonatal trypsin

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Approximately15%of patients with Cystic Fibrosis (CF) present with intestinal obstruction in immediatiate postnatal period secondary to ispissation of tenacious meconium in the ileum. This is characteristic finding: we report a case CF newborn presenting "apple peel atresia".

C. was a 1910 g male infant born after 32 week twin pregnancy. His fetal ultrasound showed bowel obstruction. He had bilious vomiting at birth: at surgery jejunum atresia type IV with distal ileal loop wraping an Anomalous Mesenteric Axis (Apple Peel Disease) were found. Resection of about 20 cm of distal loop and 5 cm of proximal loop was performed. A second operation was necessary for a perforation in an ectasic loop, seven days after. The child presented early onset of cholestasis and Rx, at 21 day of age, showed lobar atelectasis of right upper lobe. Analysis of specific mutations showed the presence of F508del and 541delC; sweat test was abnormal: Cl 96 mEq/L. Neonatal serum trypsine (IRT) level was normal. The other two brothers had normal sweat test.

The term Apple Peel atresia describes a particular type of intestinal atresia charachterized by an occlusion of the jejunum, reabsorption of the mesentery and a wrapping of a large part of the small intestine around a thin vascular stalk consisting of the left branch of the ileocolic artery. Although 10–15% of infants with jejunoileal atresia are know to have CF, Apple Peel disease has reported in association with Cf only in two cases.

In this report we point out the strict association between Cf and Apple Peel disease and underline, as reported for meconium ileus, normal values of IRT at birth, suggesting that also in this intestinal features, sweat test should be performed.

376 Sweat test results on a suspected cystic fibrosis population with CF mutation analysis and the frequency of borderline results

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Sweat test analysis were evaluated from suspected population of Ege Region of Turkey and CF mutation analysis performed from cases with high sweat test results. Sweat test was expressed as mmol/L (equivalent NaCl)conductivity. 1904 patients were included in the study, sweat test results of 31 patients were found to be higher than 80 mmol/L NaCl conductivity and 16 patients' test results were between 60–80 mmol/L. 53 patients were undergone to CF mutation analysis. 2183AA>A, 2789 + 5G>A, G542X, N1303K, W1282, R347P, ΔF508 mutations were seen frequently in all those patients. No mutations were detected in some of them although with very high sweat test results. Surprisingly, ΔF508 mutation was not the most common mutation as it was considered to be. There was a significant negative correlation between sweat test result and body mass index with an F value of 0.026. Borderline cases were seen in adults, a very frequent circumstance in normal adult populations of both sexes and two of them were the fathers of CF diagnosed patients.

Normal adult values, though very frequently falling into the borderline range for children, never reached the abnormal level. In fact, there is no borderline area for adults. However, if an adult has a frankly CF sweat conductivity level, confirmed by repeat sweat tests, they must be considered positive. The results also show that the body mass index could be a very useful marker for borderline cases. It was abnormally low in CF subjects. Therefore, it might be valuable in the exclusion of CF in cases with no mutations detected.